

Health Care Provider Fact Sheet

Disease Name

Short-chain acyl-CoA dehydrogenase deficiency

Acronym

SCADD or SCAD

Disease Classification

Fatty Acid Oxidation Disorder

Variants

Yes

Variant name

Late-onset with chronic myopathy

Symptom onset

Neonatal - but very variable; may be asymptomatic.

Symptoms

Neonatal - failure to thrive, hypotonia, metabolic acidosis, seizures and developmental delay.

Natural history

Developmental delay, hypotonia and muscle weakness have been observed, but the vast **without treatment** majority of patients detected via MS/MS newborn screening have been entirely asymptomatic.

Natural history with treatment

The efficacy of treatment is unknown.

Treatment

Carnitine supplementation, restriction of dietary fat. A few patients have shown improvements on riboflavin supplements.

Other

Acute fatty liver of pregnancy and HELLP syndrome have been reported as maternal complications in pregnancy, but may be coincidental.

Emergency Medical treatment

See sheet from American College of Medical Genetics (attached) or for more information, go to website: <http://www.acmg.net/StaticContent/ACT/C4.pdf>

Physical phenotype Inheritance

None reported
Autosomal recessive

General population incidence

1:40,000 – 1:100,000

Ethnic differences

None

Population

N/A

Ethnic incidence

N/A

Enzyme location

Mitochondrial matrix in liver, muscle, fibroblasts

Enzyme Function

Missing Enzyme

Short-chain acyl-CoA dehydrogenase

Metabolite changes

Ethymalonic acid in urine, methylsuccinate and butyrylglycines in urine.

Gene

ACADS

Gene location

12q22-qter

DNA testing available

Yes - but interpretation of results is difficult.

DNA testing detail

Limited mutational hotspots and common susceptibility alleles

Prenatal testing

Enzymatic

MS/MS Profile

Elevated C4 Cbutyrylcarnitine

OMIM Link

<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=606885>

Genetests Link

www.genetests.org

Support Group

FOD Family Support Group

<http://www.fodsupport.org>

Organic Acidemia Association

<http://www.oaonews.org>

Save Babies through Screening Foundation

<http://savebabies.org>

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